Vlad Zaha, MD (00:17):

Welcome to the Hypertrophic Cardiomyopathy podcast title, The Role of Echocardiography in the Diagnosis and Management of Patients with Hypertrophic Cardiomyopathy. This is one of a series of podcasts from the American Heart Association Hypertrophic Cardiomyopathy Initiative, sponsored by Bristol Myers Squibb. My name is Vlad Zaha. I am an assistant professor at the University of Texas Southwestern Medical Center in Dallas, Texas. I'm a multi-modality advanced cardiovascular imaging specialist at the UT Southwestern Hypertrophic Cardiomyopathy Center of Excellence and medical director of the cardio-oncology program at the UT Southwestern Simmons Comprehensive Cancer Center. I have a pleasure to have three guests. Dr. William Zoghbi is the Elkins Family Distinguished Chair in Cardiac Health, chairman of the department of cardiology at the Houston Methodist DeBakey Heart and Vascular Center and professor of medicine at Weill Cornell Medical College and the Institute of Academic Medicine. He is past president of the American Society of Echocardiography and the American College of Cardiology.

Vlad Zaha, MD (01:24):

And Dr. Said Alsidawi, he's an assistant professor of medicine at Mayo Clinic Alix School of Medicine with primary practice at Mayo Clinic, Scottsdale, Arizona. He did advanced echo training imaging at Mayo Clinic in Rochester with the main area of expertise in hypertrophic cardiomyopathy and valvular heart diseases.

Vlad Zaha, MD (01:44):

And we have with us a patient representative, Cris Iaboni, who will be discussing with us several questions. So, we'll start with the first question about the significance and role of echocardiography in diagnosis of hypertrophic cardiomyopathy directed to Dr. Zoghbi.

William Zoghbi MD (02:04):

Echocardiography or cardiac ultrasound is really the first modality to evaluate the heart. And particularly in hypertrophic cardiomyopathy. And the beauty of ultrasound is certainly it's non-invasive. It can be done frequently, but as a first diagnosis, it is essential for us to take a look at the heart muscle since hypertrophic cardiomyopathy is a muscle disease. And how is the thickness of this heart? If there is a question or there is a relation of an individual is affected with their siblings or their relatives. So for screening and for looking at structure of the heart, it is really the first modality. And the importance of ultrasound in this situation is take a look at heart function, heart thickness, where is hypertrophic cardiomyopathy involving this heart? At times, it involves the whole heart. Most commonly, actually, it involve the septum, which is the area between the two big chambers of the heart. And at times, there are issues with some obstruction within this heart, in the area of the mitral valve. At times, it can occur in the middle of this heart at the ventricle.

William Zoghbi MD (03:18):

And at times, you may have a little pouch or an abnormality of contraction of the apex of the heart. So therefore, the role of ultrasound is to take a look at this whole heart structure, its function. What are the associated abnormalities that you may see in hypertrophic cardiomyopathy? And where is the area of abnormality in the heart that has been affected?

Vlad Zaha, MD (03:41):

Thank you. That is excellent. And turning to Cris, now, I was wondering if you would be able to tell us about your diagnosis. Was that made by echo?

Cristina Iaboni (03:53):

Yes, it was. It was the primary way that I understood what was going on in my heart and how to address it and deal with it.

Vlad Zaha, MD (04:04):

And do you recall at the time when they did the test, whether you were resting or whether you had a stress test?

Cristina Iaboni (04:11):

The first time back in 2016, I had a stress test associated with it.

Vlad Zaha, MD (04:18):

And was it a regular echocardiogram that identified your diagnosis or was there any special test that they needed to do?

Cristina Iaboni (04:27):

It was just the echocardiogram.

Vlad Zaha, MD (04:29):

Okay. Cristina, thank you for your answer. I put a couple different questions was maybe to highlight, but maybe Dr. Alsidawi will give us a little bit more detail. Maybe that was not something you realized during your echocardiogram that they found the diagnosis immediately, that you did not need necessarily the stress test on that follow up study when they put the diagnosis.

Cristina Iaboni (04:50):

So, when I did the echo in 2021 with Dr. Alsidawi, he stopped the stress piece. We never actually got to it because the LVOT was so high even at rest, that it didn't make any sense.

William Zoghbi MD (05:18):

Also it's important, we want to make sure we don't give the impression that stress testing is the first thing in this condition. Although, Cris, fortuitously, sounds like they found this during a stress echocardiogram. The diagnosis actually should be done at rest and in selective cases, you do a stress test for this. So, we may want to address that a little bit in the transition towards Dr. Alsidawi.

Vlad Zaha, MD (05:45):

Yeah, I think that's a good point because I was trying to surface that from the experience, but maybe it's not obvious for the patients, right? So that's an important point. So, turning now to Dr. Alsidawi, I would like to explore for the role of echocardiograms in screening for hypertrophic cardiomyopathy in family members of patients with hypertrophic cardiomyopathy, especially the ones who are genetically positive and as we call them phenotypically negative in the first stage.

Said Alsidawi, MD (06:17):

I think doing genetic testing in all patients who have hypertrophic cardiomyopathy is a very important thing because it makes the screening of their family members much easier. So, patients who are test positive for a pathologic or a likely pathologic gene, we can go and do cascade testing for their first-degree relatives. And those who test positive, need frequent follow up. Those who test negative are done and can be saved from having any further testing. We have to keep in mind that only 50% of patients who undergo genetic testing end up being positive. So, we have a group of patients who have negative genes and then, we do the same. We treat their family members because we don't know if they have the gene or not as if they are genetically positive. So, the way we currently follow patients who do not have a family gene or who are genetically positive, is essentially the same.

Said Alsidawi, MD (07:03):

We do frequent echocardiogram and EKGs on them. The time at initiation is essentially with any symptoms or at the age of 12 year old. And between the age of 12 and 20 year old, we do echocardiograms and EKGs every one to two years. After the age of 20, we spread out the screening again with EKGs and echoes to every three to five years. Except for professional athletes, we continue to screen them more frequently at times. Doing the echocardiogram, needs very careful attention to details in patients with family members of hypertrophic cardiomyopathy patients. The cutoff, we use is a little bit lower for the diagnosis. We use a cutoff of 13 millimeters to give the patient the diagnosis because of their family history or if they're genetically positive. Also, we should pay attention to other details such as abnormalities in the papillary muscle, abnormalities in the submitral apparatus. Also, the apical basal muscle septal bands.

Said Alsidawi, MD (07:59):

All these features suggest that this is a patient who potentially can go ahead and have a full blown hypertrophic cardiomyopathy in the future. We don't treat them differently, but these are features that to pay attention to while we're screening these patients. Most of us stop screening at the age of 60 as the incidence of hypertrophic cardiomyopathy after that at age islow.

Vlad Zaha, MD (08:21):

Excellent. Thank you. Cris, what was the experience in your family regarding echocardiographic screening for hypertrophic cardiomyopathy?

Cristina Iaboni (08:32):

So after I met with Dr. Alsidawi in 2021, we talked about the genetic screening and I did go through that process. However, nothing came out of that, that was significant or that my family members could be screened for. My two brothers and my two sons have had echocardiograms and none of them showed any sign of HCM at this point. It's all good. And they'll continue to be screened as Dr. Alsidawi has suggested.

Vlad Zaha, MD (09:04):

Thank you. Dr. Zoghbi, would you like to comment for us a little bit more on the role of echocardiograms, both at rest and maybe stress for understanding and managing the symptoms of patients with hypertrophic cardiomyopathy?

William Zoghbi MD (09:20):

This is a very important question. I think people need to know that many patients actually with hypertrophic cardiomyopathy depending on their stage may be asymptomatic. And when symptoms occur, it's a variety of symptoms. Most common is that of shortness of breath, usually during exertion, less during rest, and at times, some discomfort during exertion, maximal exertion in the chest. And the third could be any arrhythmias that you may have palpitations, arrhythmias, atrial fibrillation. People hear a lot about atrial fibrillation. And the third one, obviously the one that we want to avoid or at least combat is the more serious ones, usually they may present with sudden death. So, this is the one that we want to make sure that we avoid or treat or at least have in our mind for the high risk. So for the symptoms, the echocardiogram tells us a lot about, one, heart function, is the ventricle still pumping the way it should be? Usually, it's more hyperdynamic.

William Zoghbi MD (10:24):

Two, for people who have some shortness of breath, is there obstruction somewhere? And this thicker myocardium that is very vigorous. And most of the time, if there is obstruction, it is at the level of the mitral valve or what we call the left ventricular outflow, where the blood is being ejected from the heart itself. At times, there is an obstruction within the cavity itself, almost in the middle of it, or it could be towards the tip. So, we look for those. The third that we look for in trying to understand symptoms is how much abnormality of relaxation does this heart have. With a thicker heart, the heart doesn't relax as well after every beat. And if there are a lot of relaxation abnormalities, the pressure in diastole when the heart relaxes, this is not the blood pressure that we're talking about in the arm, the pressure within the heart, maybe elevated and then, certainly can get worse during exertion.

William Zoghbi MD (11:21):

So, we always look at that with ultrasound and Doppler examination within the ultrasound and last but not least is what is the pulmonary pressure, the pressure within the lung that is being subjected in that particular patient to understand the whole situation. Now, at times, patients may not have an obstruction and they're still very symptomatic, mostly from shortness of breath. And this is the situation where you do some stress testing, either with exercise or with dobutamine, which is simulation, just to see if obstruction is simulated during an exertion as opposed to rest. And we know that very well, that the degree of obstruction, if somebody has an obstruction is very variable. It depends how much fluid you have on the body. If you're dehydrated, if you took a diuretic, if you take as certain medication, it is quite variable. And I think the physicians as well as the patients need to know that.

Vlad Zaha, MD (12:19):

Thank you. Dr. Alsidawi, would you like to comment further about the role of echocardiography in this case, going beyond diagnosis and thinking about guidance during surgical or procedural interventions for hypertrophic cardiomyopathy?

Said Alsidawi, MD (12:36):

Absolutely. When patients fail medical management, the next therapy is septal reduction therapy. And here we're talking about the specific phenotype of obstructive hypertrophic cardiomyopathy. The role of the echocardiography is extremely important to define the anatomy of the heart and then, guide which procedure the patient should get and what exactly needs to be done during each procedure. So as you know now, we have two ways of doing septal reduction therapy, whether it's surgical myectomy or alcohol septal ablation. The role of the echocardiographer is to define the thickest segment of the heart and to try to describe the mechanism of obstruction, you can have multiple level of obstruction, not only

in the LVOT, it could be intracavitary. It could be towards the apex. So, the echocardiographer has to help by defining where the level of obstruction is trying to help decide which procedure would work best for the patient.

Said Alsidawi, MD (13:25):

We need to understand the anatomy of the papillary muscle and how much they're contributing to the obstruction. We have to understand if there's anything wrong with the mitral valve as well because long term, there could be mitral valve abnormalities. Depending on these things, we can recommend to the patient, surgery versus alcohol septal ablation. When it comes to surgery, during the surgery, our institution, echocardiographers perform the intra-op TEE. We do very careful assessment of the anatomy. Again, the role of the papillary muscle, we study the mitral valve very carefully, so we can help the surgeons make decisions about what exactly needs to be done during these surgeries. During alcohol septal ablations, we define what exactly was supplied by the septal perforator selected for the procedure by injecting contrast beyond the balloon in the septal perforator and trying to define the area that's going to be affected by the alcohol septal ablation.

Said Alsidawi, MD (14:18):

After surgery and after alcohol septal ablation, the echocardiographer should look for any possible complications, ventricular septal defects, or any other complication that potentially might have come from the procedure itself. So, I think echocardiography plays a big role in the planning during the procedure and after the procedure for septal reduction therapy .

Vlad Zaha, MD (14:37):

Thank you. Cris, so after you had the echocardiogram and you had the diagnosis placed of hypertrophic cardiomyopathy, do you recall the discussion about the treatment options and then, any follow-up echocardiograms after you had your treatment?

Cristina Iaboni (14:55):

The conversations that Dr. Alsidawi and I had around the best course of action, we discussed the alcohol ablation. We discussed septal myectomy to address the symptoms that I had and given where the thickening of the septum was and the papillary muscle affecting the valve, it was very clear that alcohol ablation just was not an option or it would not be the best option. So, we talked about septal myectomy. We talked about my level of symptoms, my quality of life, all of those types of things. It was clear that either now or at some time in the future, so myectomy was the right thing to do. We did talk a number of times actually because of my concerns, open heart surgery is a big deal, but the way to address my symptoms and my quality of life, it brought me to surgery. I don't believe we did a follow-up echo, but I do recall that when we got to the echo part, Dr. Alsidawi decided not to do any stress component to it because of my condition.

Vlad Zaha, MD (16:07):

Excellent. You brought a few really important points on understanding how the structure of the heart was changed and how the structure of the heart was then linked to the treatment options. And also regarding the stage of the hypertrophic cardiomyopathy that was raising concerns about limiting stressful tests. Dr. Zoghbi, thinking about the risk stratification for patients with hypertrophic cardiomyopathy. What are your important points that you consider in discussions with patients and with colleagues?

William Zoghbi MD (16:47):

This is very important. I think this is also what patients think about and said, what is my life is going to look like? What kind of risk do I have down the line having been diagnosed with the disease and the risk varies depending on the phenotype of how is this disease affecting the heart itself. And the way I think about risk is, there is risk of heart failure, which is not necessarily in the traditional fashion of where the heart really fails, but the heart failure with a preserved function, basically, patients are more short of breath than usual, particularly with some physical activity. Two other risk, one is sudden death, which is very obviously concerning in this entity. And the third one is a risk of atrial fibrillation and therefore, need for blood thinners in such an individual. So you have to think in totality for the various scenarios that a patient may have.

William Zoghbi MD (17:41):

So from a heart failure perspective, if we can delay or take away some of that obstruction that Cris had, and I know she had surgery for that, that decreases the event of heart failure down the line, that can also at times be managed medically. So, it depends. So, it's not everything is surgical intervention. So, the physician will talk with their patient and decide just like she decided for what to do as to what medical therapy would be best because the echocardiogram, since we're focusing on this here helps you, the physician, and the patient know how well is this therapy? What is the pressure like? So that's one. Two, regarding heart failure. What are the pressures within this heart that is not relaxing as well? Do we need additional medications to bring this pressure down? And therefore, risk stratify. The third one, obviously, which is important is the risk of sudden death.

William Zoghbi MD (18:39):

And that risk of sudden death puts together, if you will, various parameters of that. One is how thick is this heart? At times, if the thickness of the heart is above two and a half, three centimeters, and you could do something surgically for it, yes. And if not, you have to think of an ICD, which is obviously an internal defibrillator to prevent ventricular tachycardia and sudden death. Patients who have arrhythmias, repeated ventricular tachycardias, patients who have low blood pressure when they exert themselves. So, you have to discuss all these factors, put it together with the echocardiogram, what you see, and therefore, a good discussion between the individual and their healthcare provider to make a decision as what to go, to decrease those risks going forward, heart failure, atrial fibrillation, and sudden cardiac death.

Vlad Zaha, MD (19:34):

Excellent. That was a great summary as well as a detailed presentation of those key points. Dr. Alsidawi thinking about the long-term follow up for some patients and their families, what do you think are the points that we need to keep in mind in terms of the utilization of echocardiography?

Said Alsidawi, MD (19:57):

Dr. Zoghbi mentioned a very important point, which is surgery is saved for patients who are already maximized on medical management and failed medical management. As Cris mentioned, we really try to maximize beta-blocker, calcium-channel blocker first. Unfortunately, without any significant improvement in symptoms or her gradients in the heart, that's why we went the surgical route. The main reason we went the surgical route was because she had a very anomalous papillary muscle causing significant obstruction. So, I just wanted to clarify this point and that helped with the decision. Now, patients who have hypertrophic cardiomyopathy, we usually follow up with them on a yearly basis,

assuming they're stable. Every year, we do an echocardiogram for multiple reasons trying to see if there are any changes in their wall thickness. If they have developed any aneurysms or pouches in the heart or to check their ejection fraction.

Said Alsidawi, MD (20:49):

So essentially for risk stratification, because that has to be done every year. The patient doesn't need an ICD this year doesn't mean he doesn't need it this year. So, we have to reevaluate the whole situation on a yearly basis. Patients who already had surgery, we need to follow up also with the same frequency, trying to see, did they develop any systolic cardiomyopathy? Are they going into a new stage of hypertrophic cardiomyopathy as we call burnout hypertrophic cardiomyopathy, which requires a more advanced heart failure therapy down the road. So, we keep yearly follow up with all patients with hypertrophic cardiomyopathy, whether they underwent surgical or procedural septal reduction or not on a yearly basis, mainly to reevaluate risk stratification again and then, make sure they're not proceeding to other stage of the disease as well.

Vlad Zaha, MD (21:37):

Excellent. Thank you so much. And Cris, thinking about all the testing that had to be happening during your diagnosis and follow ups, can you tell us a little bit about how was that affecting you in terms of time and maybe financial burden as to getting through all those tests?

Cristina Iaboni (21:58):

It certainly takes time, but obviously when you're talking about something as serious as this, you make the time. The financial burden is significant. I'm very fortunate in that I have great insurance coverage, but if I didn't, I know that for every test with my provider, every echo is $5,000 deposit that not everybody can afford that. And it's unfortunate because the level of care that I believe I received was a direct result of the echo and a direct result of Dr. Alsidawi who is very well versed in echo and was able to explain all of that to me. I don't know, in the past, I had echos and I had diagnosis, but it was never really explained. I think the two things have to go hand-in-hand, the ability to get an echo, whether it's financially or otherwise and having somebody explain it really, really well.

Vlad Zaha, MD (22:58):

Okay. Thank you. I think we went through a lot of key points here. And just to summarize the main lines of our conversation, we covered diagnosis, screening for the family members, the management of symptoms, procedural guidance, follow up, and the potential burden of testing as far as time and finances for the patient. So now, I would like to turn to the group again for your final thoughts and maybe some take home messages for our colleagues and for their patients. So, Dr. Alsidawi.

Said Alsidawi, MD (23:35):

Sure. I just want to emphasize two quick points here. Now, we have center of excellence for hypertrophic cardiomyopathy. And when we say center of excellence, that means excellence at all levels, imaging, treatment management, medical management, and then surgical management. I think whenever a patient is diagnosed with this condition, it's never a bad idea to refer them for an opinion at a center of excellence, just to have experts review the case and make sure that the patient is getting the best treatment possible. I mean, the patient has the right to know what's available. And just the fact that a center doesn't offer certain treatment, I think we shouldn't hold that treatment from the patient. I think echocardiographer should pay close attentions to the diagnosis. I think we're still under

diagnosing hypertrophic cardiomyopathy, because sometimes patient are asymptomatic, not coming in or because we're missing the diagnosis on echo. Paying close attention to details, utilizing contrast, strain imaging, all possible advanced imaging, trying to reach the right diagnosis, explain it to the patient and making sure the patient knows all the available options for them is extremely important.

Said Alsidawi, MD (24:38):

And second thing, I want to emphasize the importance of the 2020 hypertrophic cardiomyopathy guidelines and the importance of shared decision making. Sometimes, we see a lot of gray zone cases where ICD maybe, maybe not indicated. Maybe the case is surgical, maybe not. I think the patient decision and understanding of the disease is extremely important here. And we should move away from the approach of telling the patient what should be done and involve them in the decision making. And I think the current guidelines play a big role emphasizing this point.

Vlad Zaha, MD (25:09):

Excellent. And as the patient's experience was emphasized by Dr. Alsidawi. Cris, I would like to have maybe your final points that may complement that.

Cristina Iaboni (25:20):

I completely agree with Dr. Alsidawi. My experience has been from 2009 till today that if the knowledge that the medical professional has isn't clearly communicated to the patient in a manner that that patient is involved and understands the disease, the potential progression, the treatment options, et cetera, the patient could easily just ignore it and move on. As I did in 2009 and again in 2016. So having that patient involved means explaining in a manner that they understand what's going on and what could happen in the future.

Vlad Zaha, MD (26:05):

Thank you. To conclude, Dr. Zoghbi, do you have, have any remarks that you'd like to share with us?

William Zoghbi MD (26:11):

Certainly. I love this format because a knowledgeable patient is probably a most powerful patient for themselves because it appeases them in a way and makes them work closely with their physicians, so that communication is crucial between the team. I cannot overemphasize, a message, I would say of hope for patients because nowadays as opposed to 15, 20 years ago, our knowledge of the disease, what we can bring for to patients to feel better, not only feel better from a physical point of view, but also from a mental point of view, because of the concerns of sudden cardiac death down the line with ICD therapy, medical therapy, surgical, catheter based therapies. There are so many things that we could do. Yes, we haven't answered everything, but we are certainly much better off nowadays than before.

William Zoghbi MD (27:10):

So therefore, if somebody has a disease like this and is wondering what to do, what's the best approach? This is where you get an opinion from, there are multiple centers for hypertrophic cardiomyopathy to see where's the role of echo. I know we're emphasizing that today. Was there of all the other therapies, when do you use an MRI, to put them all together, but echo for the long run certainly is the diagnostic modality to be able to follow patients, treat them, see if they have. And my last message actually to patients is that as you grow with the disease and we want you to live with this disease and live well with the disease is don't forget about other things that you need to take care of,

some physical activity, blood pressure, diabetes, cholesterol control, because we want to live as long as possible and doing well.

Vlad Zaha, MD (28:07):

Oh, excellent. Those are very wise comments that will be valued by everybody that's in our community and our patients. So, thank you all for participating in this podcast with me today. A lot of important points about echocardiography used in diagnosing and managing hypertrophic cardiomyopathy. This podcast is part of the American Heart Association Hypertrophic Cardiomyopathy Initiative, sponsored by Bristol Myers Squibb. And in closing, I'd like to remind everyone listening to encourage your patients to play an active role in their medical care by advocating for themselves and their family members. To get additional information, please visit American Heart Association's hypertrophic cardiomyopathy website for more education. Thank you.